

ABSTRACT

METHOD FOR DETERMINING THE PRESENCE OR ABSENCE OF DIFFERENT ANEUPLOIDIES IN A SAMPLE

The invention provides a method for determining copy number variations (CNV) of a sequence of interest in a test sample that comprises a mixture of nucleic acids that are known or are suspected to differ in the amount of one or more sequence of interest. The method comprises a statistical approach that accounts for accrued variability stemming from process-related, interchromosomal and inter-sequencing variability. The method is applicable to determining CNV of any fetal aneuploidy, and CNVs known or suspected to be associated with a variety of medical conditions. CNV that can be determined according to the present method include trisomies and monosomies of any one or more of chromosomes 1-22, X and Y, other chromosomal polysomies, and deletions and/or duplications of segments of any one or more of the chromosomes, which can be detected by sequencing only once the nucleic acids of a test sample. Any aneuploidy can be determined from sequencing information that is obtained by sequencing only once the nucleic acids of a test sample.

[Figure 19]

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[Figure 19]